The DMRF is leaving no stone unturned in the pursuit of a cure.
What is Dystonia?
Dystonia is a disorder that affects the nervous system. Improper signaling from the brain causes muscles to contract and twist involuntarily. Dystonia can affect a single body area or multiple muscle groups. There are several forms of dystonia, and dozens of diseases and conditions include dystonia as a significant symptom. For more information visit: http://www.dystonia-foundation.org

On the Cover:
For 35 years, the DMRF has been a leader in stimulating research to better understand the impact of dystonia on the brain and nervous system. Dr. Devin Binder spoke about the latest surgical techniques to treat dystonia at a recent symposium in California. See page 7 to read about the research projects the DMRF is funding this year.

Partial support of the Dystonia Dialogue is provided by educational grants from Allergan, Inc., The Medtronic Foundation, and Merz Pharmaceuticals.
Foundation Update

Dear Friends,

The leadership of the Dystonia Medical Research Foundation (DMRF) believes the greatest service we can provide the dystonia community is a focused, unrelenting push for improved therapies and a cure. Though progress can never come fast enough, we are leaving no stone unturned in search of new research funding sources and scientific collaborations that bring us closer to our ultimate goal. We operate as an agent of the hopes and priorities you convey in your emails, phone calls, social network postings, and letters. The DMRF works for you, and we promise to work tirelessly to improve the lives of families impacted by all forms of dystonia.

An important recent event, made possible by the Dystonia Advocacy Network (DAN), is the inclusion of dystonia as a condition eligible for study in the Department of Defense’s (DOD) Congressionally Directed Medical Research Program for the second year in a row. The DOD had never supported dystonia investigations before FY 2010, so this is a previously untapped source of precious federal funding. In 2010, two important investigations were funded, and we remain optimistic that dystonia projects will be funded in 2011. As a member of and administrative center for the DAN, the DMRF is part of the effort to keep dystonia on the exclusive list of disorders approved for the DOD research program. The DMRF was invited to provide volunteers to testify before the Senate Defense Appropriations Subcommittee to encourage the DOD to continue investing in dystonia research. We are grateful to Dee Linde of Portland, Oregon who traveled to Washington, DC in June to testify for this purpose. See page 17 for information about Dee’s experience.

Our efforts to improve the lives of individuals with dystonia requires the DMRF to, as a member of DAN, survey the legislative environment for laws and regulations that impede or advance our progress. In this issue of the Dystonia Dialogue you will read about this year’s Dystonia Advocacy Day, and we encourage you to learn more about all the DAN’s efforts at http://www.dystonia-advocacy.org

As long as individuals are diagnosed with dystonia, clearly there is an urgent need for resources to support them now. The DMRF is committed to providing support groups, educational meetings and symposiums, webinars, online social networking forums, a website and materials that contain timely and accurate information, and to serve as a point of contact for your questions. The daily challenges of living with dystonia can be all-consuming, and the DMRF leadership wants you to know that you have a whole team working on your behalf to provide the dystonia community with a better future. To those of you who are able to join us in these efforts through your donations, volunteerism, and advocacy, we are profoundly grateful. For as much as the DMRF works for you, we also cannot do it without you.

We appreciate your support. Thank you for being a part of the DMRF. If there is anything we can do to serve you better, please do not hesitate to contact us.

Sincerely,

Art Kessler
President

Janet L. Hieshetter
Executive Director
Survey Says...

The DMRF posted an anonymous online survey entitled, How Do You Cope? Many thanks to all who participated. Here are some of the results.

**Top 5 things that help you cope:**
- Rest & Sleep – 69%
- Maintaining a Positive Attitude – 62%
- Humor – 54%
- Prescribed Medications – 54%
- Support from Family – 53%

79% said pain is a part of their dystonia experience.
69% said depression is a part of their dystonia experience.
76% said anxiety is a part of their dystonia experience.

What has surprised you about living with dystonia?

“How socially isolating it can be, and how freeing it can be to reach out to others.”

“How it totally impacts every aspect of living.”

“You find out who your true friends are.”

“The exhaustion.”

“The difficulties it brings not only to you, but your family also.”

“How much I’ve been able to do in my life since my diagnosis.”

“I am not good at predicting a ‘good day’ or ‘bad day.’ Dystonia seems to have a mind of its own!”

“That so many people have never heard of dystonia.”

“When I explain it to people, they have been very understanding.”

“That my own family seems to think I’m faking for attention.”

“How you adapt to progressive disability.”

---

**Upcoming Events**

<table>
<thead>
<tr>
<th>Event</th>
<th>Date</th>
<th>Location</th>
</tr>
</thead>
<tbody>
<tr>
<td>3rd Annual Minnesota Dystonia Golf Tournament</td>
<td>Aug 19, 2011</td>
<td>Hastings, Minnesota</td>
</tr>
<tr>
<td>4th Annual Dystonia Golf Classic</td>
<td>Oct 3, 2011</td>
<td>Germantown, Maryland</td>
</tr>
<tr>
<td>6th Annual New Jersey Dogs4Dystonia</td>
<td>Oct 9, 2011</td>
<td>Freehold, New Jersey</td>
</tr>
<tr>
<td>5th International Dystonia Symposium</td>
<td>Oct 20–22, 2011</td>
<td>Barcelona, Spain</td>
</tr>
<tr>
<td>2M Walk</td>
<td>Oct 27, 2011</td>
<td>Santa Clarita, California</td>
</tr>
<tr>
<td>New Jersey Tricky Tray</td>
<td>Oct 28–29, 2011</td>
<td>West Paterson, New Jersey</td>
</tr>
<tr>
<td>Northeast Dystonia Symposium</td>
<td>Oct 28–29, 2011</td>
<td>Needham, Massachusetts</td>
</tr>
<tr>
<td>2M Walk</td>
<td>Oct 16, 2011</td>
<td>Santa Clarita, California</td>
</tr>
<tr>
<td>5th Annual New Jersey Dogs4Dystonia</td>
<td>Oct 16, 2011</td>
<td>Santa Clarita, California</td>
</tr>
<tr>
<td>5th International Dystonia Symposium</td>
<td>Oct 16, 2011</td>
<td>Santa Clarita, California</td>
</tr>
<tr>
<td>New Jersey Tricky Tray</td>
<td>Oct 28–29, 2011</td>
<td>West Paterson, New Jersey</td>
</tr>
<tr>
<td>Northeast Dystonia Symposium</td>
<td>Oct 28–29, 2011</td>
<td>Needham, Massachusetts</td>
</tr>
</tbody>
</table>

See the calendar at www.dystonia-foundation.org for a complete list of events as dates are confirmed.

---

Dystonia made me a lazy chef, but I enjoy food prepared by others all the more!

Who said this? Find out at www.dystonia-foundation.org/faces_of_dystonia
**Dystonia 101**

Dystonia can be a confusing disorder to understand. It never hurts to brush up on the basics:

- Dystonia is a neurological disorder. It affects the nervous system’s ability to control voluntary muscle movements.
- Dystonia does not affect smooth muscles, such as the heart.
- There are many forms of dystonia. It can affect a single body area (focal dystonia) or multiple muscle groups (segmental and generalized dystonia).
- Dystonia can exist on its own, or be a symptom of another neurological or metabolic disorder.
- In primary dystonia, the affected person has no other neurological symptoms and the dystonia is known or suspected to be genetic.
- In secondary dystonia, the symptoms can be attributed to other diseases/conditions or an insult to the nervous system such as physical trauma or drug-exposure.
- Each case of dystonia is classified by the age symptoms started, whether it can be classified as primary or secondary, the body parts affected, and the presence or absence of other disorders.
- Treatment options include oral medications, botulinum neurotoxin injections, surgery, and less invasive methods such as physical and occupational therapy, and relaxation practices.
- While stress does not cause dystonia, symptoms may worsen in stressful situations.

For more information, visit [http://www.dystonia-foundation.org](http://www.dystonia-foundation.org)
Drug Target Development Contract Enters Phase 2
DMRF & Tyler’s Hope Join Forces to Pursue Drug Targets

The DMRF and Tyler’s Hope for a Dystonia Cure are co-funding the second phase of a contract with BioFocus, a European biotechnology company, to pursue targets for the discovery and development of new dystonia drugs. The DMRF launched the first phase of the project in 2009. Phase one created an assay to identify genes and proteins that modify torsinA, the protein associated with the DYT1 dystonia gene mutation. The project has now entered phase two, and the assay will be used for robotic screening.

ARTICLE AT A GLANCE
• The DMRF is working with BioFocus to identify new drug targets for dystonia.
• Tyler’s Hope joined forces with the DMRF to help fund the second phase of this project.
• The project is a unique, large-scale effort to identify novel therapeutic targets for DYT1 dystonia and hopefully other dystonias.

The genes and proteins identified in this project will be potential drug targets and may be used in subsequent drug discovery programs to develop therapeutics to alleviate dystonia symptoms.

“No one else is doing this kind of work,” says DMRF Vice President of Science Deborah Kilpatrick, PhD. “If we don’t pursue this, who else will? We have an obligation to push the research forward to the next level.” Dr. Kilpatrick lives with cervical dystonia which causes painful muscle contractions and spasms in the neck and shoulder muscles.

The joint DMRF-Tyler’s Hope project with BioFocus is a first step to rationally design novel treatments for DYT1 and possibly other dystonias. The history of dystonia treatment is full of examples of therapeutics first intended for one form of dystonia that prove beneficial for multiple forms (examples include botulinum neurotoxin and deep brain stimulation), so there is hope that novel, mechanism-based approaches for DYT1 dystonia treatment could prove universally useful for other dystonias.

A project of this significance would not be possible without the preceding decades of dystonia research or without a dedicated full-time science staff. The project is jointly managed by David Fisher, PhD of BioFocus and Jan Teller, PhD of the DMRF with the help of dystonia experts D. Cristopher Bragg, PhD and Xandra Breakefield, PhD of Massachusetts General Hospital, Phyllis Hanson, MD, PhD of Washington University School of Medicine, and Andrew Singleton, PhD of the National Institute on Aging.

Dystonia Included in DOD’s Research Program
Investigators Can Apply for Dystonia Research Funding

The DMRF is pleased to announce that dystonia has been included in the FY 2011 Department of Defense (DOD) Congressionally Directed, Peer-Reviewed Medical Research Program. This program provides support for military health-related research. Dystonia has been included for the second consecutive year. The amount appropriated for the entire program in FY 2011 is $50 million.

The inclusion of dystonia as an eligible condition was made possible through the work of the Dystonia Advocacy Network (DAN) which is comprised of the Benign Essential Blepharospasm Research Foundation, DySTonia, Inc., the Dystonia Medical Research Foundation, the National Spasmodic Dysphonia Association, and the National Spasmodic Torticollis Association. Interested investigators can view the Program Announcement and guidelines for applicants issued by the DOD at: https://cdmrp.org/files/2011/prmrp/ttda_pa.pdf. Dystonia is included in the list of Congressionally Directed Topic Areas.

Congratulations to our DAN advocates who worked tirelessly on this issue! See page 20 to read about efforts to keep dystonia on the list of conditions eligible for study in the DOD research program.
The ultimate goal of the DMRF Science Program is to support the discovery of improved therapies and a cure. To achieve this goal, the DMRF is dedicated to stimulating the field of dystonia research and supporting the collaborations and projects necessary to accelerate progress.

The DMRF is also devoted to attracting young, talented investigators who are becoming the next generation of leaders in dystonia research. In many cases, DMRF grant and fellowship awards are intended to help investigators gather enough preliminary data to make them eligible for much larger grants from agencies such as the National Institutes of Health.

The DMRF is proud to announce this year’s exciting research projects, some of which are continuing from 2010. Each of these projects is meaningful because it addresses one or more of the core directions necessary to advance the field. These core directions include furthering our fundamental understanding of what dystonia is, uncovering the mechanisms in the nervous system that lead to symptoms, creating models of dystonia to use in experiments, and discovering targets for new and improved therapeutics designed specifically to treat dystonia.

Congratulations to this year’s award recipients and infinite thanks to our supporters for making this research funding possible.

GRANTS & CONTRACTS
Identification of Novel Drug Targets for DYT1 dystonia
BioFocus, DPhil
Leiden, The Netherlands
This project is the first step toward the rational design of dystonia therapeutics by identifying new potential drug targets.

A Yeast Expression System to Uncover Fundamental Aspects of TorsinA Function
Jeffrey Brodsky, PhD
University of Pittsburgh
Pittsburgh, PA
Michal Zolkiewski, PhD
Kansas State University
Manhattan, KS
To clarify the role of torsinA, this project explores molecular differences between normal and mutated torsinA produced in common brewer’s yeast. This project unites the expertise of two established investigators and their labs.

Characterization of Neuronal and Non-Neuronal Forms of THAP1 Protein
Michelle Ehrlich, MD
Mount Sinai School of Medicine
New York, NY
Mutations in the THAP1 gene are associated with DYT6 dystonia, a form of early onset dystonia, and may be related to the more common focal dystonias as well. Dr. Ehrlich plans to learn more about the protein associated with this gene and the relationship between the mutated protein and symptoms of dystonia.

How Does the DYT1 Dystonia Mutation Alter TorsinA Function?
Rose Goodchild, PhD
University of Tennessee
Knoxville, TN
Dr. Goodchild has discovered that the mutated form of the dystonia protein torsinA interacts with recently discovered proteins LAP1 and LULL1. She is exploring the role of these new proteins in normal torsinA function.

Transgenic Rat Models for DYT1 Dystonia
Kathrin Grundmann, MD
University of Tübingen
Tübingen, Germany
The goal of this project is to develop the first rat model of DYT1 dystonia which will offer new opportunities to study this form of the disorder.

Abundance and Cellular Localization of Wildtype and Mutant SGCE in Human Skin Fibroblasts and Induced Pluripotent Stem Cell-derived Neurons
Anne Grünwald, PhD
University of Lübeck
Lübeck, Germany
Myoclonus dystonia is caused by mutations in the epsilon-sarcoglycan (SGCE) gene in about 25% of cases. This study seeks to clarify why only a percentage of individuals who inherit the gene manifest symptoms and the differences between the proteins associated with normal and mutant SGCE. This may help clarify penetrance patterns for additional dystonias.
Activity-dependent Synaptic Plasticity in Globus Pallidus of Dystonia Patients
William Hutchison, PhD
Toronto Western Hospital
Toronto, Ontario
Dr. Hutchison is investigating whether electrical stimulation can be used to ‘recalibrate’ brain networks and allow the brain to ‘relearn’ a movement task without producing dystonia symptoms. This work is a step toward developing treatments to target these mechanisms.

Dystonia Gene Discovery Through Autozygosity Mapping and Massively Parallel Sequencing
Michael Kruer, MD
University of South Dakota
Sioux Falls, SD
Henry Houlden, MD, PhD
University College London
London, UK
This project seeks to identify previously unknown genetic causes of dystonia using novel comprehensive genetic screening. The findings have the potential to significantly advance our understanding of dystonia at the molecular level and identify new mechanisms of dystonia.

Neurophysiological Study of Myoclonus Dystonia
Sabine Meunier, MD, PhD
National Institute of Health and Medical Research (INSERM)
Paris, France
John Rothwell, PhD
University College London
London, UK
Although the protein associated with the gene responsible for myoclonus dystonia is very common, it is not clear why mutations in this gene cause the jerks and spasms that are so apparent in patients. This study uses new methods of brain stimulation to learn more about how symptoms arise. The findings will provide clues toward new treatments.

Understanding the Role of the Cerebellum in Dystonia
Vikram Shakkottai, MD
University of Michigan
Ann Arbor, MI
Dr. Shakkottai is attempting to clarify the role of an area of the brain that controls balance and movement, the cerebellum, in dystonia. If the cerebellum is shown to play a pivotal role in dystonia, this may suggest new strategies for treatment.

Pharmacological Assessment of Muscarinic Receptor Antagonist Potency and Selectivity
Vastra, Ltd.
Corbridge, Northumberland UK
This project involves testing of drug and drug-like compounds that interact with specific receptors in the brain. These compounds are thought to have an impact on the way the brain controls movements. This study will provide basic information about the properties of these compounds and the likelihood of their success as a potential treatment option for patients with dystonia.

Clinical and Genetic Studies of African American Patients with Dystonia
Zbigniew Wszolek, MD
Mayo Clinic Jacksonville
Jacksonville, FL
Dr. Wszolek, in partnership with Dr. Mark LeDoux, is studying the genetics of several African American families impacted by dystonia, hoping to identify new gene(s) associated with adult onset, focal dystonia. This study will expand what is known about the genetics of focal dystonias and is providing important outreach to dystonia-affected African American families in Southern Georgia and Northern Florida.
Characterization of Substrate Interactions with TorsinA
Li Zhu, PhD, Chinese Academy of Sciences, Beijing, China

The DYT1 dystonia mutation appears to disrupt the ability of torsinA to play a role in regulating interactions with other proteins within the cell. In this project, the interaction between torsinA and other proteins will be characterized, and undiscovered torsinA partner proteins may be identified. This work will help to further our understanding of the function of torsinA.

FELLOWSHIPS
Optogenetic Manipulation of Striatal Fast Spiking Interneurons in vivo
Daniel Leventhal, MD, PhD, University of Michigan, Ann Arbor, MI

This research focuses on one cell type, called striatal fast spiking interneurons (FSIs), which appear to be associated with various forms of dystonia by applying a revolutionary optogenetic technology. The experiments will provide important data on the role of FSIs in normal and abnormal states.

A Novel Nonhuman Model to Probe Postural Control and Plasticity
Simon Overduin, PhD, University of California, Berkeley, CA

This study proposes that dystonia is a malfunction of the brain’s ability to coordinate sequences of movements and postures. This research introduces a new reversible primate model of dystonia, one that may allow scientists to explore the brain’s ability to plan movements in the normal state and to rehabilitate muscle control in the diseased state.

Neuroanatomical Abnormalities in DYT1 Dystonia
Chang-hyun Song, DVM, Emory University, Atlanta, GA

Exactly which region of the brain is responsible for the abnormal signals associated with dystonia symptoms is a matter of intense recent debate in the neuroscience community. To better understand how symptoms originate, Dr. Song is exploring the brains of DYT1 mutant mice for suspected anatomical defects in three main brain regions currently believed to be involved in dystonia.

Defining the Parameters of TorsinA Function in Neural Development
Lauren Tanabe, PhD, University of Michigan, Ann Arbor, MI

Dr. Tanabe is using a new mouse model to pinpoint when and how the dystonia protein torsinA is functioning during the formation of the nervous system in embryonic development. She is also exploring the role of a related protein, torsinB, as possibly protecting the nervous system from the effects of mutant torsinA.

Glossary

**DYT** – A designation given to genes associated with dystonia. At this time there are more than 20 dystonia genes and associations.

**DYT1** – The gene associated with early onset generalized dystonia is the DYT1 gene. A specific mutation in this gene can lead to dystonia.

**DYT6** – A gene associated with early onset primary dystonia affecting the face and neck areas of the body.

**DYT11** – A form of dystonia characterized by mutations in the epsilon-sarcoglycan (SGCE) gene, also known as myoclonus dystonia. This form is characterized by a combination of rapid, jerking muscle contractions and dystonia.

**DYT16** – A gene associated with early onset dystonia-parkinsonism. Also called the PACT or PRKRA gene.

**Mutation** – A change in the genetic material of a living thing. The result of a mutation is a “mutated” or “mutant” gene and its corresponding protein. For example, the DYT6 dystonia gene mutation results in a mutant form of the protein THAP1.

**Penetrance** – The percentage of people with a specific disease gene mutation who actually develop symptoms of the disease. For example, approximately 30% of people who inherit the DYT1 dystonia mutation develop symptoms. This means the remaining 70% of people with the gene mutation never develop dystonia.

**TorsinA** – The name of the protein associated with the DYT1 gene and early onset generalized dystonia. The DYT1 gene mutation causes changes in the torsinA protein and how it functions.
Oasis in the Desert
New Mexico Support Group is Rare Resource for Affected Individuals

In 1994 Alfred “A.J.” Deguio experienced his first brush with dystonia. While driving on a New Mexico highway, his eyes inexplicably closed and he could not open them. “Thank God it only lasted a few seconds,” he recalls. “I was frightened to death.” Understandably rattled, A.J. drove the short distance remaining to his home and wondered what on earth had just happened. The next morning, dystonia visited again.

He recounts how his “whole face just grabbed, tightened up under my neck. My eyes were blinking. I absolutely could not tolerate any lights, television. My eye area was sensitive to touch and would trigger twitches and rapid blinking. I couldn’t read even a page in a book—not even one line at times!”

When the symptoms did not relent for 10 days, A.J. consulted an optometrist. The optometrist referred him to an ophthalmologist who promptly concluded, “I think you should see a neurologist.” A.J. ultimately found himself at the office of movement disorder specialist Neal Hermanowicz, MD and was diagnosed with benign essential blepharospasm, a focal dystonia of the eye lid and brow muscles. He was 62 years old, having recently retired from the aviation industry.

A.J. threw himself into learning as much as possible about blepharospasm and the other dystonia disorders. Fifteen years ago he started a blepharospasm support group and was struck by the number of individuals with other forms of dystonia who contacted him because there were no other local dystonia support groups to be found. A.J. welcomed them all and the group quickly evolved into a support resource for all forms of dystonia. “All dystonia people are invited,” he says. Resources for individuals with dystonia are limited in New Mexico, and movement disorder specialists are few.

The New Mexico Dystonia Support Group meets every other month on Saturdays. Some meetings feature guest speakers, and some meetings focus on group discussion. A.J. respects the needs of the attendees and conducts the group as democratically as possible. His priority is to bring people together: “Support groups are necessary. There is no way you can get the same satisfaction without the face to face contact.”

"Newcomers need to know what they are dealing with, what the future may look like,” he continues. “This is a chronic disorder, so you’re probably stuck with it, but it’s also a functional disorder—not degenerative—so a cure is possible.”

For many years, A.J. underwent botulinum neurotoxin therapy. However, what seems to help him cope best was simply learning to accommodate the dystonia. “Now my right eye is closed most of the time. My left eye is open 75% of the time. The symptoms are improving each year. I can watch TV, tolerate light, and read the newspaper. I know my limits, and I can do most everything I need. I just can’t push my luck.”

To search for your nearest DMRF support group, go to http://www.dystonia-foundation.org and look for the link under “Living with Dystonia.” Or contact the DMRF office at 800-377-DYST (3978) or dystonia@dystonia-foundation.org
Go, Gigi, Go!
Girl Runs Race for Her Mom

Six-year-old Gigi Kenney’s mom Yanett has dystonia. When she asked her mom, “When are you going to get better?” her mom explained that dystonia is not the kind of illness that usually goes away. Gigi then asked, “What would you do if you get cured?” Her mother said she would take a walk to the Lafayette Reservoir, where she used to take Gigi all the time before dystonia made it too hard to walk. So Gigi made a decision. She decided to run in the Twilight 5K race to raise money for dystonia research. She would train for the race by running at the Lafayette Reservoir after school twice a week with her nanny.

The Lamorinda Weekly newspaper wrote a story about Gigi and her plans for the race. Friends and businesses donated to Gigi’s cause. She raised $500!

“She finished her 5K race in about 45 minutes—with a small detour to run and hug her mom before crossing the finish line,” reported Yanett. “We are so proud of her!”

The DMRF is also extremely proud of Gigi, who is one of the youngest Dystance4Dystonia volunteers. What an inspiring young lady!

What’s Up Docs?
Daughter & Mom Help Educate Doctors

Melissa Rentfrow and mom Kathy Rentfrow of Maryland have staffed a dystonia awareness booth at local doctors’ conferences for several years. They focus on educating pediatric neurologists on the forms of dystonia that affect children. Great job!

Awesomeness for $500, Alex!
DMRF Member Wins Kids Jeopardy

B.D. Schwarz of California celebrated his 13th birthday on July 6 by watching himself win on the TV game show “Jeopardy!” His family and friends were there to celebrate, and many others tuned in to cheer him on.

B.D. was selected to be one of only 15 children featured on Jeopardy Kids Week, July 5–9, 2011. He said, “Going on Jeopardy was an enriching experience. I met many wonderful people who were just as nice as they appeared to be on TV. Alex Trebek is such a nice guy. I’m still not sure this all wasn’t a dream.”

Many congratulations to B.D. for this special achievement. He is an inspiration to other kids with dystonia and adults too!
PEOPLE ON THE MOVE

One of the most important strengths of the DMRF is the amazing nationwide team of volunteers in local communities. The DMRF is extremely fortunate to benefit from the hard work of individuals and families across the country who promote awareness and fundraise. Every effort and volunteer makes a difference! Sincere thanks to all our grassroots volunteers and supporters.

Early this year board member and Network Liaison Sandy Weil hosted the annual Joan Rabbiner Memorial Brunch in Boca Raton, Florida to benefit the DMRF. Vice President of Public Policy Rosalie Lewis delivered the keynote address and shared her family’s experience with dystonia. Sandy is among the DMRF’s supreme champions of fundraising, working tirelessly in honor of her son Dr. Rob Weil who developed generalized dystonia as a child.

In January, Melinda and Larry Wolthuis of Delton, Michigan hosted a dystonia awareness booth at two local health and wellness fairs. Melinda was diagnosed with dystonia in 2009. An avid birdwatcher, Melinda also raised funds for the DMRF by making and selling Valentine’s Day seed cakes—heart shaped clusters of bird food to help feed our feathered friends.

Every Sunday during football season, Michelle Kraft and the “Buckeye Packer Backers” meet at On Tap Pub in Columbus, Ohio to watch the Green Bay Packers. Each week they have a 50/50 raffle, earmarking 50% of all proceeds to charity. In 2011, the group selected the DMRF as a designated charity, raising $1,000! Michelle and several of her family members are affected by myoclonus dystonia.

Earlier in the year, the Support and Advocacy Group of San Diego County welcomed back member Alex John whose struggle with dystonia and journey through deep brain stimulation (DBS) surgery were documented in several episodes of the Emmy-winning television program, The Doctors. Paul Fowler presented Alex with an “O” Award named in memory of musician Otha Young, who was integral in organizing several benefit concerts featuring recording artist Juice Newton to benefit the DMRF. Alex is doing very well following her DBS, so much so that she traveled to the meeting on the back of a motorcycle, which is something dystonia had prevented her from doing for many years.

Pat Brogan, who appeared in the PBS film Twisted, is registered to compete in his first ever triathlon, the Dewey Beach Sprint Triathlon on September 17, 2011. Ten years ago, while training for what was supposed to be his first triathlon, Pat was hit by a car. He suffered a brain contusion which led to dystonia. Pat is determined to “offer hope to anyone who suffers from a disability or any obstacle in life to prove that anything can be accomplished through hard work and perseverance.” Go, Pat!

In March, Dystance4Dystonia volunteer Julie Zaia competed in the Shamrock Marathon in Virginia Beach, Virginia in honor of her sister Stephanie Zaia, who has generalized dystonia. Julie collected sponsorships from family and friends, and finished the race in 3 hours – 35 minutes. Congratulations, Julie!
Krista Richter competed in the grueling Leona Divide 50 Mile Ultramarathon in Lake Hughes, California on behalf of the DMRF this spring. Amazing!

Thanks go out to the Tabile family of Sal & Joe’s Restaurant in Maple Shade, New Jersey and the family and friends of Guido and Rita Battaglini who supported the Dine for Dystonia Event in March at the restaurant. This one-night event raised $2,450! Pictured are Bob Kamuca, Rita Battaglini, Sal Tabile, and Phil Tabile. Not pictured are Joe Tabile and Guido Battaglini, who has cervical dystonia.

Dystonia-affected musician and Awareness Ambassador Billy McLaughlin partnered with the Support and Advocacy Group of San Diego County to host a concert to benefit the DMRF. D.C. Hathaway, also a dystonia-affected musician, was the opening act. CBS News sent a film crew to shoot the concert and interview Billy for a segment of The Early Show which aired in April. “To have national coverage of the story of Billy’s triumph over dystonia is a goal we have all been striving for,” remarked organizer Nancy Dennison.

In May Mark Tsai, Regional Account Manager from US WorldMeds, spoke about the dystonia therapy Myobloc® to the Support and Advocacy Group of San Diego County led by Martha Murphy. In June Martha and Bette George hosted a booth at Summer Healthcare Saturday in San Diego to promote awareness and appeared on a local TV station covering the event.

Joey Tehle of Fargo, Idaho completed her very first 5K walk to promote dystonia awareness. Joey’s dystonia began at age 10 and eroded her ability to speak, walk, and write. At one time, she could not have walked 30 feet, let alone over three miles. Her story attracted the interest of a local TV affiliate and resulted in a story about this remarkable accomplishment. Joey was supported during the race by her uncle Gary Hanson. Congratulations, Joey!

The Los Angeles Support Group hosted its first meeting under the new leadership of Von Cashman and Janis Eiler at Cedars-Sinai Medical Center. Many thanks to these new leaders for their time and effort to serve the Los Angeles dystonia community.

Sandra Calvert-Nathans and Robert Nathans once again hosted a neighborhood yard sale to benefit the DMRF. They also sold several items online to boost proceeds, raising $3,500! The Nathans support the DMRF in honor of their children, Jack and Lydia, who are both diagnosed.

In April, the DMRF hosted the Seattle Dystonia Educational Forum featuring dystonia experts Susie Ro, MD; Ronald Young, MD; and Patrick Hogan, DO, MD. Many thanks to all the attendees for participating and to our speakers for lending their time and expertise.

Paula and Don Gates and their fabulous committee once again put the fun in fund-raising by hosting the annual “Return to Margaritaville” event. This Jimmy Buffett-inspired dinner dance raised an amazing $21,000! Infinite thanks to the Gates and their dedicated family and friends.

The 34th Rochester Institute of Technology Spring Juggle-In took place in April. For the last 11 years the Juggle-In has focused on raising awareness and funds for research to cure dystonia, inspired by Rosalie and Richard Lewis and their four sons, three of who are diagnosed. The RIT club has raised over $20,000 for the DMRF. The festival was founded by Greg Moss and is now coordinated by Jeff Peden.
This spring Patricia Bergeron and the Verville Family organized the 6th Annual Hands for Movement Freedom benefit piano recital in Vermont. Approximately 30 pianists participated, all of who are students of Patricia Bergeron. The event raised over $300!

Sally Presby of Omaha, Nebraska organized and hosted a “Fat Tuesday Fundraiser” featuring a special performance by vocalist Carmelita de la Guardia. The event raised nearly $1,000 and promoted much-needed awareness. Terrific work!

Sculptor Melinda Eames, inspired by childhood friend and fellow artist Antonella Gosselin, founded the annual Women Who Weld exhibit each spring in Omaha, Nebraska to benefit the DMRF. This year’s show, entitled “Walk with Fire” featured 12 area artists, each of who donated a sculpture to be auctioned to benefit the DMRF. Melinda explains, “Antonella inspires us to create art that tells a story of passion and strength.”

Leader of the Dystonia Support Group of Alabama, Pat Wyatt, once again met with Governor Robert Bentley who proclaimed the week of June 5–11 as Alabama Dystonia Awareness Week. Pat is pictured with the Governor plus husband Bill Wyatt and grandson Eric Arrowood. Pat also appeared in the Shelby County Reporter during Dystonia Awareness Week. Excellent work!

In May Hunter Webster hosted the 2nd Annual Dogs4Dystonia Dog Walk in Fairfield Park, Virginia. The event attracted a great crowd and raised important funds in support of the DMRF. Through Hunter’s efforts, the “Interstate Hotels & Resorts Gives Back” program again sponsored the walk at the Bronze level. Thank you to all of the participants and supporters who helped to make this year’s Dogs4Dystonia Dog Walk a great success!

The DMRF Chicago Chapter held their annual letter-writing campaign hosted by board members Barbara and Dennis Kessler. Support group members gather to compose personal letters inviting family, friends, and colleagues to support the DMRF. Thank you to all who participated and to the Kesslers for their leadership in this wonderful Chicago fundraising tradition.

April Arnold is a Systems Analyst within the Health Information (HIS) Department at St. Francis Hospital and Medical Center in Hartford, CT and arranged for a June fundraiser to benefit the DMRF in honor of her 16-year-old sister Amanda Arnold who has paroxysmal dystonia. The HIS Department raised $825 by collecting for "Jeans Day" every Friday, holding a bake sale, selling DMRF bracelets and donated items, and a raffle. April also made it possible for the Allscripts Enterprise Group to support the DMRF through a number of employee events.

Sabina Gall of Romania partnered with area doctors to host the country’s first known educational meeting on dystonia. She also founded a support group and has appeared in local news media to promote awareness. Inspiring work!

Which “Face of Dystonia” had to give up his career as a professional musician but is now a medical student?

Find out at http://www.dystonia-foundation.org/faces_of_dystonia
Many thanks to **Becky and Mike Cortis** and **Michelle Carrano** for hosting Northampton, Massachusetts’ first Dystance4Dystonia Walk to Raise Awareness. The DMRF also thanks the volunteers, Best Buy, and additional sponsors who helped to make this event such a success.

The Greater Dallas Dystonia/Dysphonia Support Group led by **Peggy Akin** hosted a meeting in June featuring guest speakers **John Desaloms, MD** of the Presbyterian Hospital of Dallas and one of his most recent deep brain stimulation patients **Amy Behar**. **Allergan** provided lunch and several representatives from **Medtronic** were also in attendance. The meeting attracted the group’s largest crowd to date, totaling over 50 attendees.

**Memoirs of a Cheetah**

**DMRF Member Dolores September of Portland, Oregon shares an excerpt from her unpublished work, Memoirs of a Cheetah, which chronicles her experience with dystonia to promote greater awareness. Below is a brief peek into the first chapter, when seven-year-old Dolores begins to experience her first symptoms of dystonia.**

“Before long, my left hand twitched from morning till night and could no longer be trusted to carry anything fragile or hot. I began to stammer. My parents just looked at each other. A few months later when I walked down the aisle of St. Michael’s to receive my First Holy Communion, I appeared like any other girl in white, except for my head, which tilted to the left as though I was straining to hear a quiet question. No one mentioned this after church as friends and I congratulated each other, and family members embraced me. And no one mentioned it that evening at our celebration dinner as my dad raised his class of Scotch high and toasted me with tears in his eyes and confidence on his lips that his daughter was healthy and smart and had only good fortune ahead of her.

“The next day I heard my Aunt Wanda tell Mom, ‘I wouldn’t worry. Maybe Dolores just pulled a muscle.’ However, as the weeks and months passed, all muscles acquired a tough will of their own, yanking me this way and that. My left ear inched toward my left shoulder. My spine curved. My right hip jutted out. Aunt Wanda, a professional seamstress, now made my dresses four inches longer in back to cover my thighs when I stood or moved. Perspiration from the effort to walk was profuse and democratic enough to leave my clothes sweat-soaked in all seasons. I felt controlled by a sadistic puppeteer.”


**Students Support DMRF for Charity Day**

Every year, students at Middlesex Valley Elementary in Rushville, New York select two charities to focus on for Charity Day. The DMRF was one of the charities chosen this year. On May 14 the students hosted a bake sale, car wash, and other activities to collect donations. They raised almost $500 for the DMRF! Student Ashley Smith, who has a family member with dystonia, won the charity poster contest with her poster about dystonia awareness. Many thanks to Ashley and all the kids at Middlesex Valley Elementary for their hard work and support!
Back in the 1970s I was attending a Naval Engineering Facilities East Coast Symposium on Oil Spills in Virginia Beach. During the symposium I was asked out of the blue to address the attendees on logistical support during oil spills. I was rather in shock at this request and nervous as I approached the stage and stood behind the podium. After talking for over an hour I stepped down and one of the engineers asked me if I had “slept wrong” the night before because I acted as though I had a “kink in my neck.” I wasn’t aware of my posture at the podium and passed it off to being nervous.

During the next few years I started to experience periods when my neck would pull over and be painful whenever I got upset or mad about something. The Navy medical facilities were busy and doctors were in short supply, so they put me out on CHAMPUS (authorized to use civilian doctors). I retired and went to work for the Navy as a contractor. My visits to various doctors resulted in the conclusion that I was having muscle spasms due to an injury I had sustained many years earlier causing severe whip lash. They wanted to give me pain medicine which I refused because I wouldn’t have been able to perform my specialized duties if medicated.

One evening I was really having a time with my neck, and I went to the emergency room at the Naval Hospital. I was seen by a young Navy doctor on duty who happened to be right out of medical school. That was my lucky day. He checked me out, read my medical records, and said he had an idea what my problem might be but wanted me to go out on CHAMPUS to see a neurologist to confirm his evaluation. I was diagnosed with cervical dystonia. The neurologist asked me to visit another neurologist in Boston who specialized in dystonia and, if the diagnosis was correct, to receive medical treatment. The Navy doctor had written what he thought may be my problem on a piece of paper and asked me let him know if he was right. I called the Navy doctor and told him he had guessed right and that I was going to see a specialist in Boston.

I went to the specialist in Boston who confirmed the diagnosis and explained treatment options for dystonia. I let him inject me with botulinum neurotoxin, but refused the oral medication that he wanted to prescribe because my exposures in the military could contribute to the possibility of certain side effects. The botulinum neurotoxin injections I received helped to relieve the symptoms. Later I chose to see a different doctor in New York who I had met through some friends in a support group.

After a period of time the botulinum neurotoxin injections had less effect on my dystonia. Since I was able to reduce the effects of dystonia temporarily myself through breathing exercises I learned while in the Navy to deal with stress, I stopped seeking medical assistance for my problem.

It has been around 20 years since I sought any medical help for my dystonia. I have learned to deal with my neck issues, the resulting back issues, and a compressed lung. I am now fully retired. My wife and I moved back to our home state of Maine where the winters are long and cold. Our winters affect my cervical dystonia, but I returned home to enjoy all the things I missed while serving my country which includes the outdoors in Maine. I spend most winter days out in the cold. I trap for additional income, hunt all winter for the varied game available, ice fish, and cut wood to heat our home. I have not and do not take any medication for dystonia, except for some ibuprofen after a particularly bad day so I can sleep. The bad days I have are few and far between. I still
I am amazed after all these years that people I meet are afraid or embarrassed to ask why my neck pulls over to the side. I have not let dystonia rule my life. I truly believe if a person is positive-minded, keeps their body as flexible and strong as possible because of physical work, learns a method (like my breathing exercise) that works for them to relax, and doesn’t pay attention to those who pity you, they can deal with cervical dystonia. If they are embarrassed because of it, look around at all the other people in the world who are worse off. I only have to visualize my comrades in the military who were injured and burned to realize how truly blessed I am.

I have a loving wife who I’ve been with since our high school days and who has never coddled me, and I wouldn’t want her to. Get over feeling sorry for yourself and fight with all that’s within you. No battle was ever won without an all-out fight.

I will be forever grateful to that young Navy doctor who figured out what my problem was and pointed me in the right direction. I will never forget his name—“Lt. Muller”—and if he stayed in the service I am grateful as the Navy needed people like him. If he got out and practiced on the outside, the civilian world got a fine doctor. Where ever he is today I hope he sees this and knows he is well appreciated and loved.

On Wednesday, June 22, dystonia advocate Dee Linde of Portland, Oregon appeared before the Senate Defense Appropriations Subcommittee to testify in support of keeping dystonia as one of the “conditions eligible for study” through the Department of Defense’s (DOD) Congressionally Directed, Medical Research Program. The 2010 and 2011 Defense Appropriations Bills included dystonia on the eligible conditions list, but the list is renewed each year and being named one year is no guarantee that a condition will be named the next year. Dee was uniquely qualified to present the views of the dystonia community as both an individual affected by dystonia and a Navy veteran. Dee also served with two other DMRF members on a special Consumer Panel as part of the DOD’s dystonia research program in 2010 which included reviewing research applications and providing a patient perspective to the peer-review process.

Dee articulated to the Subcommittee the concern about dystonia among military personnel due to the connection between dystonia and traumatic brain injury, and the apparent increase in dystonia cases among veterans since the onset of the Iraq and Afghanistan wars. Chairman Daniel Inouye (D-HI) thanked Dee for her time and said the Subcommittee would “do their best” to include dystonia again as one of the conditions eligible for study through the DOD’s research program as they begin work on the 2012 Defense Appropriations Bill. Being called to testify before the Senate Defense Appropriations Subcommittee is reserved for a handful of individuals each year, and the DMRF was extremely pleased that the Subcommittee once again decided to summon a representative of the DMRF to represent the dystonia community. “It was quite an honor,” says Dee.

The webcast of the hearing including Dee’s remarks can be viewed at http://appropriations.senate.gov/webcasts.cfm. Look for the link “DOD Outside Witness Hearing” dated June 22, 2011.

Which “Face of Dystonia” coaches his 4-year-old’s t-ball team from his wheelchair?
Find out at http://www.dystonia-foundation.org/faces_of_dystonia
Taking Charge of Your Care

Taking charge of your dystonia treatment means being a proactive patient. Being a proactive patient means asking questions.

A proactive patient is one who has dedicated him/herself to living well with dystonia. You may have many questions regarding dystonia and your treatment. In order to get the most out of your medical appointments, prepare yourself for each health visit, establish a rapport with your medical team, and develop a system to take notes or identify a person to assist you in taking notes.

Your doctors and health care team work for you. Do not be intimidated or afraid to ask anything. You have a right to fully understand all of your treatment options and any potential side effects associated with your choices.

Be prepared. Do research or have a loved one do it for you. The more educated you are, the better questions you will be able to ask. Prepare a written list of questions for your doctor or nurse prior to each meeting. It can be difficult to remember each question when a lot of information is being exchanged during an office visit. Ask for clarification of any tests or procedures you do not understand.

Establish rapport. It is important from the beginning to establish a positive relationship with your physician and health care team. Keep in mind, good teams communicate well. As such, your doctor will have questions for you and likewise you will have questions for him/her too. This is part of a healthy rapport.

ARTICLE AT A GLANCE

• Taking charge of your dystonia treatment means asking questions.
• Prepare yourself for each medical appointment to make sure you get the most out of it.
• Keep your own copy of your medical record and history.
• Remember you have the right to choose your doctor, clinic, and treatment.

Focus your questions and start with the most important. Your doctor wants to answer your questions but will likely have time restraints and limitations due to many patients who require his/her time and attention. Take time to write down your questions in advance and take them with you.

Medical history. You will often be asked to recount your medical history. Write this down and always carry it with you. Information to be included: childhood illnesses, traumas and injuries, history of medication use and allergic reactions, family and medical history including cases of dystonia or other movement disorders, allergies, other medical conditions.

Build your own record. You are the center of your care team, and it only makes sense for you to have copies of your scans, x-rays, and test results. Your doctors may refer back to parts of your medical record or you may need them when seeking a second opinion. Consider keeping a health journal that includes observations about your symptoms and pain, the dates and duration of treatments, your response to treatments including side effects, and other information relevant to your dystonia and general health.

Have a record of all your medications with you. It is imperative that you keep detailed records of your medicines and complementary therapies including vitamins, supplements, and herbs.

Remember that you have choices. You have the right to choose your doctor, movement disorder center, and course of treatment.

Adapted from information from Mission Hospitals.
In spite of focal dystonia of the leg, Ben Beach of Bethesda, Maryland has competed in the Boston Marathon every spring since 1968. He holds the record for running the second most consecutive Boston Marathons. This year, at age 61, he completed the race in 4 hours – 33 minutes – 35 seconds, more than five minutes better than his time last year. Ben uses each marathon as an opportunity to promote dystonia awareness by sharing his story in the local media and The Washington Post.

Ben was previously featured in the Dystonia Dialogue in 2009 and continues to be going strong. “Aside from the dystonia, I’m pretty healthy,” he says. “I do exercises and stretches recommended by NIH [his physicians at the National Institutes of Health] and others to help my body deal with the weird gait I have. My dystonia is not painful, fortunately.”

Ben’s dystonia manifests as a mild limp that becomes more pronounced when he runs, limiting his training and making his dedication to running even more remarkable. “NIH has been experimenting with the injection sites and dosage of my Botox® since starting me on it in 2006. Finding the right formula is a challenge, as most of us have learned. I believe that the current formula is starting to pay off, and that made the marathon somewhat less difficult. Because I have so few training miles under my belt, I can count on cramping quads and a lack of energy during the second 13 miles, so I had to battle that.”

The DMRF congratulates Ben for this amazing accomplishment and for inspiring others in the dystonia community and beyond.
Dystonia Advocacy Day 2011 A Big Success

One hundred twenty enthusiastic volunteers, representing 26 states and the District of Columbia, gathered in Washington, DC on May 9–10 to advocate on behalf of the dystonia community.

The Dystonia Advocacy Network (DAN), comprised of the Benign Essential Blepharospasm Research Foundation, DySTonia, Inc., the Dystonia Medical Research Foundation, the National Spasmodic Dysphonia Association, and the National Spasmodic Torticollis Association, hosted this event which included training for new advocates, a briefing for experienced advocates, and a presentation from Steven Grossman, Deputy Executive Director of the Alliance for a Stronger Food and Drug Administration.

Dystonia advocates spent a day on Capitol Hill with over 150 meetings in Congressional offices to discuss the need for adequate funding for the National Institutes of Health (NIH), requesting a slight budget increase to ensure NIH is able to support robust research and important new initiatives like the Cures Acceleration Network. Dystonia advocates asked legislators to continue to include dystonia on the list of conditions eligible for the Department of Defense’s medical research program, in which dystonia has been included for 2010 and 2011. Dystonia advocates also discussed the need to improve access to dystonia treatments including a long-term solution to the issue of Medicare physician reimbursement rates, the need to address insurance barriers for patients who are seeking an FDA-approved Humanitarian Use Device treatment such as deep brain stimulation, and creating an approval pathway for follow-on biologic products that takes into consideration the unique safety and effectiveness concerns of people who rely on botulinum neurotoxin treatment options.

First time advocate Shellie Gray traveled from Oklahoma to participate. She experienced a few travel challenges on the way when the airline misplaced her wheelchair, but her excitement about participating was not muted. She said, “It was great to be able to tell my story not just to educate someone about dystonia, but actually to help others with dystonia. What a humbling experience to be a voice for so many!”  Shellie added, “It was an honor to represent the many, just like me, who wonder ‘Can I go to Washington, DC and make a difference in the lives of dystonia patients throughout the USA?’  I’m here to tell them, yes, you can. I witnessed it.”

The dates for next year’s Dystonia Advocacy Day are May 8–9, 2012 in Washington, DC. For more details or to become a dystonia advocate, please go to: http://www.dystonia-advocacy.org
Planned or Deferred Gifts Make a Lasting Difference

By James A. Gianelli, Esq.

Including DMRF in your estate and financial planning can allow you to fulfill your long-term philanthropic goals while realizing important benefits for yourself, your family, and/or your business.

Planned giving requires careful deliberation because it involves making a gift from your assets: cash, stocks, bonds, mutual funds, retirement accounts, life insurance policies, and real estate or personal property (such as jewelry and art). In addition, many types of planned gifts provide immediate or deferred benefits to you or your heirs, so we encourage you to speak with your professional advisors (estate planning attorneys, accountants, and financial advisors) before committing to a planned gift.

MOST POPULAR WAYS TO GIFT

1. Bequests By Will or Revocable Living Trust: Charitable bequests are effective upon your death and can be revoked or changed while you are alive. You can specify that DMRF receives a specific dollar amount, specific securities or other property, a specific percentage of your estate, or, as is typical of charitable bequests, a portion of whatever remains after other specific bequests have been satisfied. In addition, your estate (and therefore a proportionate amount of your estate tax, if applicable) is reduced by the full amount of your charitable contribution.

2. Life Insurance: By naming the DMRF as the beneficiary of a new or existing life insurance policy, you may receive a current income tax deduction equal to the policy’s cash surrender value. And if you continue to pay premiums on the policy, you may receive an additional tax deduction for those payments.

3. Retirement Assets: Qualified retirement plans, such as defined-benefit plans, profit-sharing plans, 401(k) and 403(b) plans, Keogh accounts, and individual retirement accounts (IRAs), receive favorable income tax treatment while funds remain in the plan. Upon distribution to the owner or transfer after death, however, these assets are taxed heavily under both the income and estate tax systems. You can designate the DMRF as a beneficiary of a fixed percentage of the account.

4. Bank Account in Trust: You can make a “cash bequest” to DMRF by opening or titling an existing bank or brokerage account or certificate of deposit in trust for DMRF. You retain complete control over the funds or assets in the account while you are living (and for this reason there is no current income tax deduction). Whatever remains in the account at your death is transferred automatically to DMRF without going through probate, and your estate receives a charitable deduction for the gift.

5. Charitable Trusts: As part of a well thought out estate plan under the supervision of your professional advisors, the benefits to all parties can be very rewarding:
   • Charitable trusts can minimize capital gains or inheritance taxes for you or your heirs.
   • They can provide for a sizeable deferred gift to one or more charities.
   • You may fund charitable trusts with cash, securities, real estate, retirement funds, business interests, and other assets.

While the DMRF staff is always available to assist you with charitable gift planning, it is important that you consult with your own professional advisors before making a planned or deferred gift. More information may be found at http://www.dystoniafoundation.org

James A. Gianelli received his Juris Doctorate (JD) from McGeorge School of Law in 1979 and his Masters in Law in Business and Taxation (LLM) from McGeorge School of Law in 1984. He is certified as a Specialist by the State Bar of California in Estate Planning, Trust, and Probate Law. He is a previous guest author for the Dystonia Dialogue, having shared his experience with oromandibular dystonia.
FOCUS ON X-LINKED DYSTONIA-PARKINSONISM
Meet Jesus Villanueva, MD

Dr. Villanueva is board certified in internal medicine and practiced for eight years before x-linked dystonia-parkinsonism (XDP) affected him at age 37. He now co-owns the Arizona Center for Clinical Trials, a medical research firm in Phoenix.

When and how did your symptoms begin?
I was in my third year of medical residency when my handwriting started to get smaller. My fellow residents noticed that my handwriting was hard to read. Everybody thought that it was just a quirk of being a physician. I also developed a tongue tremor and a lisp in my speech. When I first went to see a doctor, I was diagnosed with essential tremor. It was five years later that I was diagnosed by Dr. Virgilo Evidente at the Mayo Clinic with x-linked dystonia-parkinsonism, and then five years later again I tested positive for the DYT3 gene mutation.

How does XDP affect you in your daily life? Your family?
Dystonia has really affected me in my daily life. Even before my diagnosis, my gait was affecting me. I was working for a government medical agency, and my patients were complaining to my superiors that they could not understand my speech. In January of 2004, my clinic supervisor told me I had to go on terminal leave. Even though I received a small pension, my wife and the kids had to sacrifice because they could not live as well as they did in the past.

Your experience with deep brain stimulation was pretty groundbreaking—please explain.
To my knowledge, I was the first one with x-linked dystonia-parkinsonism to have the implants placed in the globus pallidus. According to my doctor, Dr. Evidente, there were reports of a Filipino man whose maternal roots came from Panay Island who had DBS [deep brain stimulation], but the electrodes were placed in the subthalamic region. As far as I know, there have been four of us who have had the DBS surgery, including my younger brother. My brother’s symptoms started when he noticed his tongue was pointing to the left. This year, he began having involuntary movements of the neck muscles. He had to have a gastric tube placed endoscopically because of the effects on his trachea. He could not eat. My brother had the first surgery of the DBS procedure done in July, and it was a success. He will have his second surgery in August. I would like to ask anybody who is reading this interview to please pray for him.

Why is it important for you to be involved with the DMRF and your additional outreach work in the Philippines?
This is the only type of dystonia which is degenerative, and I pray that a cure will be found in my lifetime. Most Filipinos are poor and even if they are in the middle class, a surgery that costs $130,000 is too expensive. The most urgent need for my countrymen who have x-linked dystonia-parkinsonism is to have medications which are difficult to access in the Philippines because they cost so much over there. I have met patients who only buy 2-4 tablets for each prescription given to them because that is all they can afford. I can only imagine how they would feel if they did not have any medication to take.
How did your symptoms begin?
I thought I was having a stroke. I had a full dystonic episode in the car with my son, who was 10. My whole body twisted and turned, I couldn't talk, it was in my tongue—I had every dystonic symptom you can think of. My son called 911. In the ER, they knew it was dystonia. They thought it was a drug reaction. They treated me like a drug addict, sent in the social worker and the psychiatrist, saying, “Now, you need to tell us what you’ve taken.” They sent me home two days before Christmas with Depakote® [valproic acid], baclofen. They pretty much kept me drugged up until I could see a movement disorder specialist in February. The movement disorder specialist diagnosed me with paroxysmal dyskinesias.

What treatment strategies help you best?
I know my triggers, weird things like going from hot to cold, stress and fatigue, travel. The episodes usually last five or 10 minutes. I get an aura before I’m going to have an episode. So I get to a safe place where I’m by myself. If people are watching it’s more stressful and it lasts longer.

What is it like to live with a form of dystonia that isn’t always obvious to other people?
When I 'look normal’ it’s very difficult for people to understand why I still need to make lifestyle changes to make sure the movements do not begin. When you look at my medical record, those last two years I was working, I was hospitalized for three or four days at the beginning of every semester and at the end of the semester—always right after the most stressful times. You work so hard to make everything happen, and then crash. I would come back from the hospital looking normal. People would talk behind my back, tell me I’m crazy. And it did bother me. I had to quit my job as a principal. I taught for a while and had to quit. The kids were wonderful. But I couldn't keep up with my work load.

What have you learned from living with paroxysmal dyskinesias?
I was in denial for years. I didn't seek help to deal with this. I thought I was a big strong person—I could handle this. When I realized I was missing out on my children's lives, I knew I couldn’t continue my negative cycle forever because the dystonia wasn't going away. At one time I could balance work and family but not anymore. So, what do I eliminate: work, the dystonia, or my family? I can't eliminate the dystonia, I'm certainly not giving up my kids, so the work had to go.

What helps you cope?
Keep a sense of humor! Sometimes every day is a challenge, but every day I look for the positive. I have a close circle of friends and very supportive family. Now I tell everyone about dystonia. I feel like it’s my job to tell everyone.
“Faces of Dystonia” is For Families

The DMRF has created a mosaic of faces and stories from the dystonia community at http://www.dystonia-foundation.org/faces_of_dystonia. Individuals of all ages, with all forms of dystonia are invited to participate by sharing a photo and completing a brief interview form. Family members are also invited to submit their stories. Submissions will be collected all year, and new profiles are posted every Tuesday.

The DMRF celebrates members of the dystonia community and invites everyone to participate in this special project. Let’s introduce the world to the dystonia community and show them who we really are.

To add your story to the “Faces of Dystonia” collection, visit http://www.dystonia-foundation.org/faces